

C99/2/rev.

Cystic Fibrosis

The Puzzle and the Promise

U.S. DEPARTMENT OF HEALTH
AND HUMAN SERVICES
Public Health Service NEWARK PUBLIC LIBRARY
National Institutes of Health

JUL 8 1981

U. S. GOVT. PUBS.
DEPOSITORY 376



HE 20.3002:

In the United States today, approximately 20,000 to 30,000 persons from infants to young adults are afflicted with cystic fibrosis.

What is it?

Cystic fibrosis (CF) is an inherited disease of children, adolescents and young adults that affects the exocrine, or externally secreting, glands of the body. These glands discharge their secretions onto the skin (sweat glands), or into organs which connect to body openings, such as the lungs and intestines, either directly or through special ducts.

In CF, the mucus-producing glands fail to produce normal, clear, free-flowing fluid. Instead, they secrete a thick, sticky mucus which tends to clog and block the ducts. This abnormal mucus accumulates in various parts of the body and interferes with vital functions, such as breathing and digestion.

Cystic fibrosis affects not only the mucus-secreting glands, but also the glands that produce sweat, as well as the salivary glands. As a result, the secretions of these glands usually contain an excessive amount of salt.

Who has CF?

In the United States today, approximately 20,000 to 30,000 persons from infants to young adults are afflicted with cystic fibrosis, a metabolic disorder of unknown cause. Equally common among males and females, CF was long considered a disease fatal in childhood. Research accomplishments in recent years, however, have increased the life expectancy of many cystic fibrosis victims well into the teens and twenties, or beyond.

CF is a congenital, noninfectious disorder inherited when both parents carry the "trait," a

CF is the most common lethal genetic disease among white children. Blacks are affected less frequently, and cystic fibrosis is rare among Oriental races.

genetic abnormality that can produce affected offspring. Although carriers can pass this trait on to their children, they themselves do not show symptoms related to cystic fibrosis. When only one parent is a carrier, the children will not inherit the disease, but have a one-in-two chance of being carriers as well. Offspring of two CF trait carriers, however, have a one-in-four chance of inheriting this disorder.

At present, there is no reliable method for identifying symptom-free carriers. Research is under way in the United States and abroad to discover a safe and effective diagnostic test for the CF trait. The development of such a test would be of great benefit to genetic counselors in predicting the likelihood for cystic fibrosis among the future offspring of prospective parents.

How prevalent is CF?

Affecting 1 in every 1,600 newborns, CF is the most common lethal genetic disease among white children. Blacks are affected less frequently, and cystic fibrosis is rare among Oriental races. Estimates indicate that there are now approximately 10,000,000 symptom-free carriers of the CF trait in the U.S. who might pass the carrier trait or the disease to their offspring. Recognized as a distinct disease only since 1938, CF is now being diagnosed with increasing frequency throughout the world.

What are its symptoms?

A disease of many disguises, cystic fibrosis often mimics other childhood disorders, complicating quick and accurate diagnosis. Generally, three major symptoms characterize CF: pancre-

Despite very large food consumption, signs of malnutrition frequently appear.

atic insufficiency, chronic respiratory disease, and a high salt content in perspiration.

In many cases, symptoms become apparent within the first few months of life. Sticky mucus secretions may interfere with breathing, digestion, or both, resulting in the infant's failure to thrive. About 10 percent of CF patients are born with an intestinal obstruction from a thick mucus-like substance that can prove fatal unless recognized promptly and corrected by surgery. Others may not develop symptoms until much later in life.

Pancreatic insufficiency

The pancreas secretes enzymes necessary for the digestive process. In most cystic fibrosis cases, however, thick mucus blocks the passageways that carry these important enzymes to the small intestine where food is broken down for use by the body, and seriously interferes with digestion. Without access to these enzymes, much of the food consumed passes through the body undigested, providing little nutritive value, and producing bulky, fatty, foul-smelling stools and abdominal pain. Because little nutritional benefit is derived from food intake, the CF victim may have a ravenous appetite. Despite very large food consumption, however, signs of malnutrition frequently appear.

Respiratory symptoms

Most cystic fibrosis patients develop chronic lung disease at some time during the course of their illness. Thick, abnormal mucus obstructs breathing passages in the lung and bronchial tree, causing enlargement of the airways, labored

A dominant feature of CF, the high concentration of salt in sweat is the most widely used diagnostic test for the disorder.

breathing and chronic cough. In time, this mucus obstruction creates a breeding ground for bacteria which leads to chronic pulmonary infections. Because these infections often become resistant to antibiotic treatment, respiratory complications cause 90 percent of all deaths from CF. Moreover, the clogged, damaged lung tissues tend to impede blood circulation, and may lead to increased blood pressure in the lung, heart strain, and eventual heart failure.

Excessive loss of salt

In cystic fibrosis, the production of perspiration with a high salt content may result in excessive salt loss during periods of increased sweating, placing patients at great risk of dehydration or heat exhaustion in the summer. Weakness and listlessness due to the loss of salt is fairly common. A dominant feature of CF, the high concentration of salt in sweat is the most widely used diagnostic test for the disorder.

CF: a profile

A cystic fibrosis patient with all the symptoms of the disease is generally undernourished, produces bulky, fatty stools, and is subject to serious respiratory infections accompanied by chronic cough and poor sleeping habits. In severe cases, additional problems may appear including clubbed fingers, bluish lips, constant bronchial infections, shortness of breath and all the complications of chronic lung disease. Primarily because of the serious lung disorder which affects most children with CF, their life span is shortened, in many cases, to under 21 years of age.

In recent years, doctors have noted that a

number of CF patients do not have all of the usual symptoms of the disorder. Many show only a partial set of complications, such as difficulty in breathing without impaired digestion, or the reverse. In fact, a few patients may look and feel perfectly well for many years. Scientists are now trying to learn why cystic fibrosis affects some persons less seriously than others.

How is CF detected?

Four major clues provide the basis for a diagnosis of cystic fibrosis:

- (1) Abnormally high salt content in perspiration
- (2) Lack of digestive enzymes in the small intestine
- (3) Chronic lung condition
- (4) Detection of CF in brothers, sisters or first cousins.



READER
FICHE

Determination of an abnormally high salt content in sweat is the simplest and most reliable test for the disorder, and generally confirms the diagnosis when typical symptoms are present. In approximately 80 percent of patients, the examination of specimens from the upper intestine will also demonstrate that pancreatic enzymes are either absent or deficient. Research is under way to develop more precise diagnostic tests for this disorder.

How is CF treated?

A prime factor in the management of the disease is a correct and early diagnosis prior to the onset of irreversible pulmonary complications. Since the severity of the respiratory condition generally determines the fate of CF victims, treatment of this symptom is of vital importance. The regimen consists of the use of a variety of antibiotics to combat lung infections, supplemented by physical therapy, and aerosol inhalation therapy or postural drainage methods to help remove mucus from the lungs.

When pancreatic insufficiency results in poor food absorption and digestion, the missing enzymes can be replaced by commercially available pancreatic extracts taken with meals. To further offset this condition, extra quantities of food — moderately restricted in fat — in a well-balanced diet are recommended. Similarly, patients take supplemental salt to compensate for its excessive loss in perspiration. This is especially important in hot weather when loss of salt may have grave consequences if left unchecked.

Today, as a result of improved diagnostic procedures and the increased use of antibiotics to control dangerous respiratory infections, the average age of survival is approximately 20 years.

What is the outlook?

Although the outlook for CF patients has improved over the years, the basic cause of this inherited metabolic disease is still not known. While clinically the disorder affects only the exocrine glands, current research suggests that a metabolic error may be present in *all* of the body's cells. The problem is further complicated by the fact that although a reliable test is available for the diagnosis of symptomatic patients, no specific method has yet been devised which will identify the symptom-free carrier of the CF trait.

Fifteen years ago, few children with cystic fibrosis lived beyond the age of 6. Today, as a result of improved diagnostic procedures and the increased use of antibiotics to control dangerous respiratory infections, the average age of survival is approximately 20 years. At the same time, the quality of life during these extended years is improving steadily.

Research in CF

With the significant advances in medical care, more CF patients are reaching an age where decisions must be faced about marriage, childbearing and careers. This fact lends renewed emphasis and importance to the work of research and treatment centers throughout the nation where efforts are under way to control and eventually to prevent this disorder.

Prominent among these centers is the National Institutes of Health (NIH) in Bethesda, Md. — the biomedical research arm of the U.S. Department of Health and Human Services.

The National Institute of Arthritis, Diabetes, Digestive and Kidney Diseases (NIADDK), a component of the NIH, bears the primary responsibility for Federally funded research programs on cystic fibrosis.

The Institute supported the program that first introduced physiotherapy in the U.S. as a standard treatment for pulmonary complications of CF. Similarly, Institute-supported investigators demonstrated the importance of broad spectrum antibiotics in controlling chronic respiratory infections, and developed effective treatment of the gastrointestinal and nutritional complications of the disorder.

Research is now aimed at identifying the basic cause and bodily effects of the disease, and at developing improved methods of prompt diagnosis, more effective treatment, detection of CF trait carriers, and eventual prevention of the disorder.

To strengthen the cystic fibrosis research effort, the NIADDK conducts an active program in its Bethesda laboratories, and supports, through research grants, investigations by many highly qualified scientists at universities and medical centers throughout the country.

In addition to the Federal research effort, a nonprofit voluntary agency, the Cystic Fibrosis Foundation at 6000 Executive Blvd., Suite 309, Rockville, Md. 20852, is dedicated to the support of research and public education on the disease.

Although cystic fibrosis offers only baffling clues to its basic defect, the clinical outlook for CF is improving, with more pieces of the research puzzle continually falling into place. While

the findings of previous years have contributed significantly to the expanding knowledge of this disorder, it is hoped that these extended avenues of research will lead to the final solution to the CF puzzle.

This pamphlet is intended to present general background information on cystic fibrosis. More detailed aspects of the disease, and the care and treatment of individual patients should be discussed with a qualified physician.

15+7



NIH Publication No. 81-223
Revised April 1981